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# POLAND SYNDROME-A CASE REPORT

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**ABSTRACT:**
Poland syndrome is a rare birth defect affecting the muscles and bones of the chest. It is characterised by the underdevelopment or complete absence of pectoralis muscle, which arises from sternum and sometimes abnormally short and webbed fingers. This condition can also affect the nearby muscles and bones, leading to various chest abnormalities. In some cases, it may also affect the heart, kidneys, liver, and digestive system. Most cases of Poland syndrome affect the right side. Boys are more commonly affected than girls. We are reporting a case of a 3 year old boy from South India with features of this syndrome.

**KEY WORDS:** Poland syndrome, Congenital Anomaly, Pectoralis major muscle, webbed fingers.

## INTRODUCTION:

Poland syndrome was first described by Sir Alfred Poland in 1841. People with Poland syndrome often have underdeveloped or missing muscles in their chest, including the pectoralis major muscle[1]. Some may have related issues like:
i) Underdeveloped or missing breasts or nipples, ii) lack of hair in the axilla, iii) Skeletal problems like missing or deformed ribs, iv) Heart, lung, or kidney defects,
v) Shortened arms or forearm bones.

This condition affects about 1 in 7,000 to 1 in 100,000 births. Males are affected 3 times more than females, and right sided presentation is twice more common than left side[2]. Children whose mothers smoked during pregnancy have more chance of developing Poland syndrome[3]. Individuals with severe symptoms such as significant chest wall abnormalities or respiratory distress may be diagnosed at birth or early childhood. Children with less severe symptoms such as mild chest deformities may be diagnosed typically between ages of 2-10 years, similar to our patient. Asymptomatic cases may not be diagnosed until adulthood, often it is an incidental finding during physical examination or imaging study.We report a case of 3 year old boy from South India. The aim of this paper is to bring awareness to health care professionals about rare cases.

## CASE REPORT:

A 3-year-old boy born of non-consanguineous marriage without any significant past history had presented to our paediatric out-patient department with complaints of short duration fever and cold.



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There was no family history of congenital anomalies. The child's developmental milestones were appropriate for age. His birth history did not suggest any significant events.

On Physical examination, his height was 91cm between (3rd-10th centile) and weight was 12kg between(3rd-10th centile).The chest showed asymmetry with depression and flattening of the right anterior chest wall region with absence of right axillary fold (figure 1). The right nipple was also displaced when compared to the left nipple. The muscle attachment on the sternum was missing on the right side. Both hands were normal. Systemic examination was normal. On Chest X-ray, the cardio-thoracic ratio was normal
(figure 2) . No other abnormality noted . Computed tomography of chest showed absence of pectoralis major and minor muscle
(figure 3).Electrocardiography and echocardiography done- were normal. Based on the findings, Poland syndrome was diagnosed. There was no need for surgical intervention in this case. Counselling to the family was given and advised for regular follow-up on outpatient basis. The patient is on regular follow-up and is doing well.

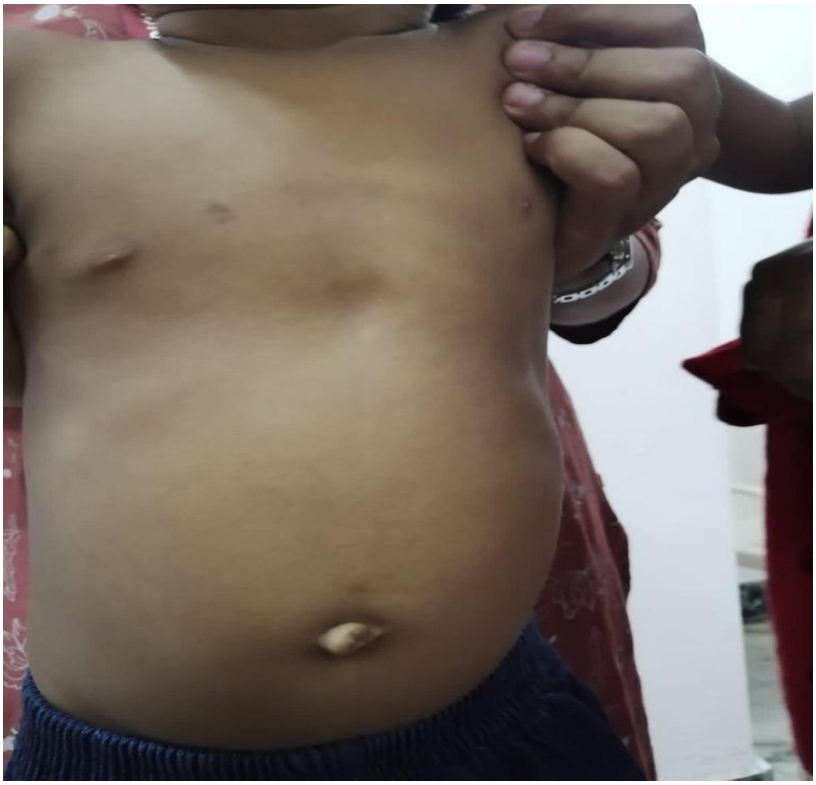

**Figure 1:** The hypotrophy of right anterior chest wall and absent pectoralis major muscle.



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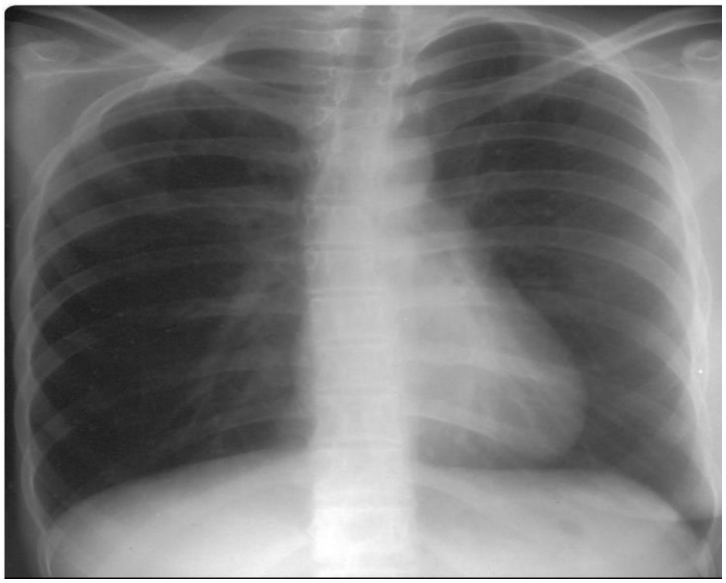

**Figure:2** Chest X-ray PA view shows normal cardio-thoracic ratio.

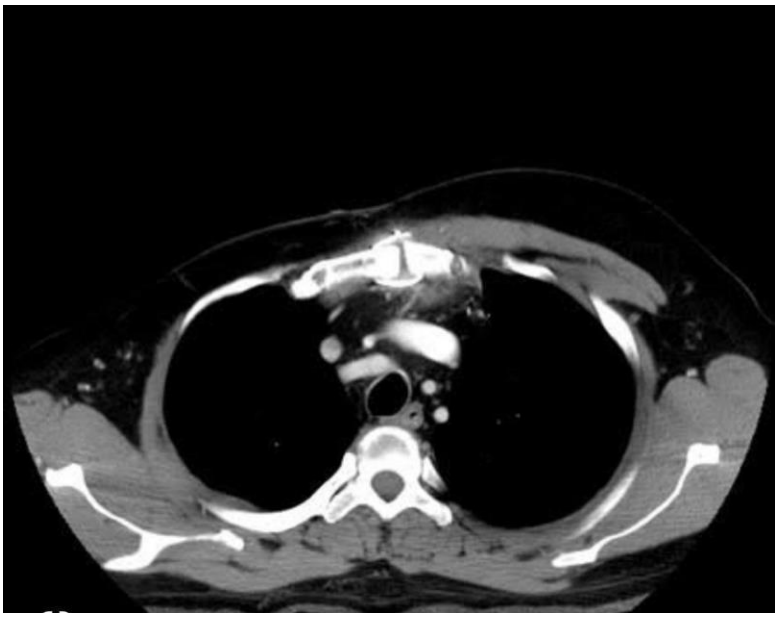

**Figure 3:** CT scan shows absence of the pectoralis major and minor muscle on right side.

## DISCUSSION

This case of Poland syndrome consists only unilateral absence of pectoralis major and minor muscle on right side without any other defects.The exact cause is unknown, and it occurs sporadically[4]. It's not inherited from parents. Research suggests that it might be related to reduced blood flow to the affected arm during fetal development, around the 6th week of

pregnancy[5].This reduced blood flow may affect the growth of the fingers and chest muscles, leading to the features of Poland syndrome. Studies have found evidence of underdeveloped blood vessels in the affected area, supporting this theory[6]. It has also been sometimes associated with skeletal, vascular (subclavian and axillary) other organ anomalies, sternal, rib abnormalities and dextrocardia.

The abnormality usually affects unilaterally and mostly the right side of the body. It can be classified into:

Type:1(minimal)- only the pectoralis muscle is absent.

Type:2( partial )- muscle defect along with upper limb or rib defect.

Type:3(complete )- muscle defect along with upper limb and rib defects.

Additional muscle involvement include lattismus dorsi, trapezius, and serratus anterior.

Treatment for Poland syndrome is based on severity of deformity. Surgery is necessary in some cases and is required to (i) Repair chest wall defects,(ii) Improve breathing,(iii) Correct chest shape, (iv)Rebuild the rib cage (v)Move the nipple-areola complex to a more natural position, (vi)Use fat injections or muscle transfers to enhance the chest area

The type of surgery used depends on the severity of the condition, the patient's age, and their gender. In some cases, bone grafts may be used to repair defects in the rib cage[7]. If the latissimus dorsi muscle is underdeveloped, the muscle from the other side of the body may be used instead. Surgery to release fused fingers (syndactyly) is usually straightforward, but it wasn't needed in this case[8]. Since our patient is asymptomatic, we are monitoring his condition without any immediate intervention.

Similar case study done in USA by Bansal et al.,of a 8 year old boy with asymmetry of the chest with depression of the anterior chest wall and displaced nipple on right side without any other signs of digital abnormality of ipsilateral side. Abduction of shoulder showed absence of sternocostal head of pectoralis major muscle[9].

Another case report by Deka et al. of a 7 month old boy from Assam showed aplasia of sternocostal, clavicular and abdominal head of pectoralis major muscle and hypoplasia of right side nipple and breast with subcutaneous fat absence on the same side without any other hand abnormalities[10].

**CONCLUSION**:

This case demonstrates the importance of thorough physical examination and radiological evaluation in diagnosing this condition. Early identification and intervention can significantly impact the child’s quality of life, enabling appropriate management and rehabilitation to optimize functional abilities. This report contributes to the existing literature on Poland syndrome,

emphasizing the variability of its presentation and need for increased awareness among health care professionals.

**PATIENT CONSENT:** obtained from the guardian.

**REFERENCES:**

1. Al Faleh K, Al Saadi M, Khalid-Bantuas S. Poland′ s syndrome with absent limb anomalies. Journal of Clinical Neonatology. 2014 Jan 1;3(1):44-6.

2.Niraula Z, Ghimire P, Pokhrel S, Magar SA, Bajracharya J, Thapa B. Incidental finding of Poland syndrome in a case of phimosis: a rare case report from Nepal. Annals of Medicine and Surgery. 2023 Mar 1;85(3):528-31.

3.Martínez-Frías ML, Czeizel AE, Rodríguez-Pinilla E, Bermejo E. Smoking during pregnancy and Poland sequence: Results of a population-based registry and a case-control registry. Teratology. 1999 Jan;59(1):35-8.

4.David TJ. Familial Poland anomaly. Journal of Medical Genetics. 1982 Aug 1;19(4):293-6.

5.Bavinck JN, Weaver DD, Opitz JM, Reynolds JF. Subclavian artery supply disruption sequence: Hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. American journal of medical genetics. 1986 Apr;23(4):903-18.

6.Beer GM, Kompatscher P, Hergan K. Poland's syndrome and vascular malformations. British journal of plastic surgery. 1996 Jan 1;49(7):482-4.

7.Alexander A, Fokin MD, Robicsek F. Poland’s syndrome revisited. Ann Thorac Surg. 2002;74(6):2218-25.

8.Braun TL, Trost JG, Pederson WC. Syndactyly release. InSeminars in Plastic Surgery 2016 Nov (Vol. 30, No. 04, pp. 162-170). Thieme Medical Publishers.

9.Bansal A, Reddy K, Dinsmore K, Gonzalez-Estrada A. Poland syndrome: a case report. Case Reports. 2017 Aug 3;2017:bcr-2017.

10.Deka D, Das BK.Poland syndrome: a case report.Int J Sci Rep2024;10(7):249-51.